ALG6 gene

ALG6, alpha-1,3-glucosyltransferase

Normal Function

The *ALG6* gene provides instructions for making an enzyme that is involved in a process called glycosylation. Glycosylation is a process by which sugar molecules (oligosaccharides) are attached to proteins and fats. Oligosaccharides are made up of many sugar molecules that are attached to one another in a stepwise process forming a complex chain. Glycosylation modifies proteins so they can perform a wider variety of functions. The enzyme produced from the *ALG6* gene transfers a simple sugar called glucose to the growing oligosaccharide. Once the correct number of sugar molecules are linked together, the oligosaccharide is attached to a protein or fat.

Health Conditions Related to Genetic Changes

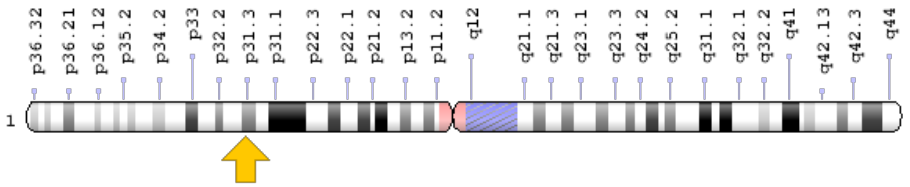
ALG6-congenital disorder of glycosylation

At least 20 mutations in the *ALG6* gene have been found to cause *ALG6*-congenital disorder of glycosylation (*ALG6*-CDG, also known as congenital disorder of glycosylation type Ic). This condition typically leads to developmental delay, vision problems, seizures, and other signs and symptoms. Mutations in the *ALG6* gene result in the production of an abnormal enzyme with reduced or no activity. A common mutation replaces the protein building block (amino acid) alanine with the amino acid valine at position 333 in the enzyme. This mutation, written as Ala333Val or A333V, results in an enzyme with reduced activity. Without a properly functioning enzyme, glycosylation cannot proceed normally, and oligosaccharides are incomplete. As a result, glycosylation is reduced or absent. The wide variety of signs and symptoms in *ALG6*-CDG are likely due to impaired glycosylation of proteins and fats that are needed for normal function in many organs and tissues, including the brain, eyes, and hormone-producing (endocrine) system.

Chromosomal Location

Cytogenetic Location: 1p31.3, which is the short (p) arm of chromosome 1 at position 31.3

Molecular Location: base pairs 63,367,590 to 63,438,562 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- asparagine-linked glycosylation 6 homolog (S. cerevisiae, alpha-1,3-glucosyltransferase)
- asparagine-linked glycosylation 6 homolog (yeast, alpha-1,3-glucosyltransferase)
- asparagine-linked glycosylation 6, alpha-1,3-glucosyltransferase homolog
- asparagine-linked glycosylation protein 6 homolog
- dol-P-Glc:Man(9)GlcNAc(2)-PP-Dol alpha-1,3-glucosyltransferase
- dolichyl-P-Glc:Man(9)GlcNAc(2)-PP-dolichol alpha- 1->3-glucosyltransferase
- dolichyl-P-Glc:Man9GlcNAc2-PP-dolichyl glucosyltransferase
- dolichyl-P-Glc:Man9GlcNAc2-PP-dolichylglucosyltransferase
- dolichyl pyrophosphate Man9GlcNAc2 alpha-1,3-glucosyltransferase
- dolichyl pyrophosphate Man9GlcNAc2 alpha-1,3-glucosyltransferase precursor
- Man(9)GlcNAc(2)-PP-Dol alpha-1,3-glucosyltransferase

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Carbohydrates Can Be Attached to Proteins to Form Glycoproteins
  https://www.ncbi.nlm.nih.gov/books/NBK22521/
- Sanford Burnham Medical Research Institute: Dr. Hudson Freeze Lab
  http://www.sbpdiscovery.org/team/hudson-freeze-phd

GeneReviews

- Congenital Disorders of N-Linked Glycosylation and Multiple Pathway Overview
  https://www.ncbi.nlm.nih.gov/books/NBK1332

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28ALG6%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- ALG6, S. CEREVISIAE, HOMOLOG OF
  http://omim.org/entry/604566

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ALG6.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ALG6%5Bgene%5D
- HGNC Gene Family: Alpha-1,3-glucosyltransferases
  http://www.genenames.org/cgi-bin/genefamilies/set/447
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=23157
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/29929
- UniProt
  http://www.uniprot.org/uniprot/Q9Y672

Sources for This Summary

- OMIM: ALG6, S. CEREVISIAE, HOMOLOG OF
  http://omim.org/entry/604566
- GeneReview: Congenital Disorders of N-Linked Glycosylation and Multiple Pathway Overview
  https://www.ncbi.nlm.nih.gov/books/NBK1332
- Grünewald S, Imbach T, Huijben K, Rubio-Gozalbo ME, Verrips A, de Klerk JB, Stroink H, de Rijk-van Andel JF, Van Hove JL, Wendel U, Matthijs G, Hennet T, Jaeken J, Wevers RA. Clinical and biochemical characteristics of congenital disorder of glycosylation type Ic, the first recognized endoplasmic reticulum defect in N-glycan synthesis. Ann Neurol. 2000 Jun;47(6):776-81.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10852543
- Imbach T, Burda P, Kuhnert P, Wevers RA, Aebi M, Berger EG, Hennet T. A mutation in the human ortholog of the Saccharomyces cerevisiae ALG6 gene causes carbohydrate-deficient glycoprotein syndrome type-Ic. Proc Natl Acad Sci U S A. 1999 Jun 8;96(12):6982-7.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10359825
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC22030/
- Westphal V, Xiao M, Kwok PY, Freeze HH. Identification of a frequent variant in ALG6, the cause of Congenital Disorder of Glycosylation-Ic. Hum Mutat. 2003 Nov;22(5):420-1.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14517965

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